

A Case of Mucopolysaccharidosis II Caused by a Novel Variant with Skin Linear Hyperpigmented Streaks along Blaschko's Lines

Viktoriia Sofronova ^{1,2,†}, Elizaveta Gurinova ^{3,†}, Diana Petukhova ², Hiroko Fukamatsu ⁴, Takenobu Yamamoto ⁴, Yumi Aoyama ⁴, Polina Golikova ^{2,3}, Gavril Moskvitin ^{2,3}, Roza Ivanova ^{2,3}, Mira Savvina ², Filipp Vasilev ², Takahito Moriwaki ¹, Seigo Terawaki ¹, Aitalina Sukhomyasova ^{2,3}, Nadezhda Maksimova ², and Takanobu Otomo ^{1,*}

¹ Department of Molecular and Genetic Medicine, Kawasaki Medical School, Kurashiki 701-0192, Japan;

² Laboratory of Molecular Medicine and Human Genetics, North-Eastern Federal University, 677013 Yakutsk, Russia

³ Medical Genetics Center, Republic Hospital No. 1 – National Center of Medicine, 677019 Yakutsk, Russia

⁴ Department of Dermatology, Kawasaki Medical School, Kurashiki 701-0192, Japan

* Correspondence: otomo@med.kawasaki-m.ac.jp; Tel.: +81-86-462-1111

† These authors contributed equally to this work

Figure S1. PCR analysis to detect presence of wild-type and deletion of exons 4-10.

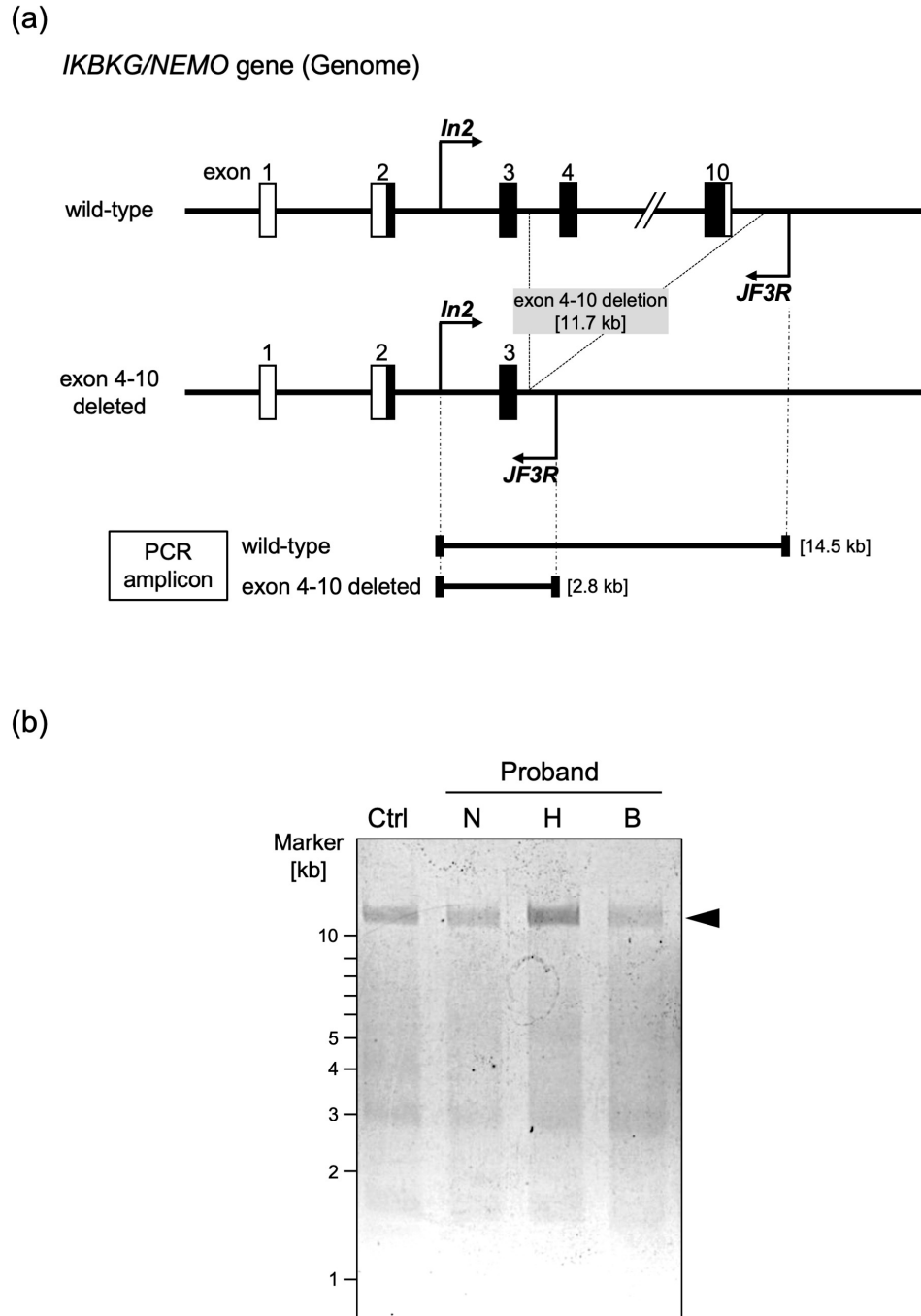


Figure S1 Schematic representation of *IKBKG/NEMO* gene and PCR results using primer set to detect exons 4-10 deletion.

(a) Structure of *IKBKG/NEMO* gene and the position of *IKBKG/NEMO* gene-specific primer set which was previously reported for the detection of exon 4-10 deletion [15]: forward primer *In2* and reverse primer *JF3R*. Expected PCR products with sizes 14.5 kb and 2.8 kb are derived from intact allele and exons 4-10 deleted allele, respectively. Coding exons are shown as black boxes. (b) Agarose gel electrophoresis of PCR products. PCR was performed using primers (*In2*/*JF3R*) with genomic DNA. Exon 4-10 deletion was not observed with this method. Ctrl: normal skin fibroblast, N: proband's skin fibroblast established from a non-pigmented skin part, H: proband's skin fibroblast established from a hyperpigmented skin lesion, B: proband's peripheral blood. Arrowhead indicates a 14.5 kb PCR amplicon corresponding to the intact allele. Marker; 1 kb ladder.